



## IRAK4 gene

interleukin 1 receptor associated kinase 4

### Normal Function

The *IRAK4* gene provides instructions for making a protein that plays an important role in innate immunity, which is the body's early, nonspecific response to foreign invaders (pathogens). The IRAK-4 protein is part of a signaling pathway that is involved in early recognition of pathogens and the initiation of inflammation to fight infection.

In particular, the IRAK-4 protein relays signals from proteins called Toll-like receptors and IL-1 receptor-related proteins. As one of the first lines of defense against infection, Toll-like receptors recognize patterns that are common to many pathogens, rather than recognizing specific pathogens, and stimulate a quick immune response. The IL-1 receptor and related proteins recognize immune system proteins called cytokines that signal the need for an immune response. The resulting signaling pathway triggers inflammation, a nonspecific immune response that helps fight infection.

### Health Conditions Related to Genetic Changes

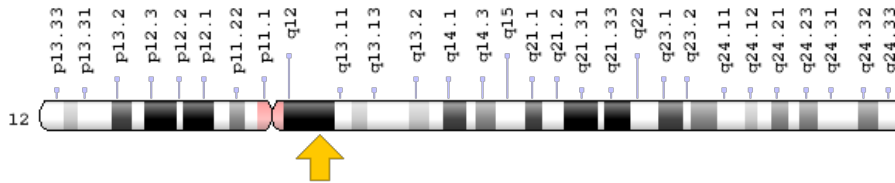
#### IRAK-4 deficiency

At least 20 mutations in the *IRAK4* gene have been identified in people with IRAK-4 deficiency, an immune system disorder that leads to recurrent invasive bacterial infections. These gene mutations lead to an abnormally short, nonfunctional IRAK-4 protein or no protein at all. The loss of functional IRAK-4 protein blocks the initiation of inflammation in response to pathogens or cytokines that would normally help fight the infections. Because the early immune response is insufficient, bacterial infections occur often and become severe and invasive.

## Chromosomal Location

Cytogenetic Location: 12q12, which is the long (q) arm of chromosome 12 at position 12

Molecular Location: base pairs 43,758,913 to 43,789,543 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- interleukin-1 receptor-associated kinase 4
- IPD1
- IRAK-4
- IRAK4\_HUMAN
- NY-REN-64
- REN64

## Additional Information & Resources

### Educational Resources

- Immunobiology: The Immune System in Health and Disease (5th Edition, 2001): A Comparison of the Drosophila and Mammalian Toll Signaling Pathways  
<https://www.ncbi.nlm.nih.gov/books/NBK27138/figure/A2372/>
- Madame Curie Bioscience Database: The Function of Toll-Like Receptors  
<https://www.ncbi.nlm.nih.gov/books/NBK6219/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28IRAK4%5BTI%5D%29+OR+%28interleukin-1+receptor-associated+kinase+4%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- INTERLEUKIN 1 RECEPTOR-ASSOCIATED KINASE 4  
<http://omim.org/entry/606883>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_IRAK4.html](http://atlasgeneticsoncology.org/Genes/GC_IRAK4.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=IRAK4%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=17967](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17967)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/51135>
- UniProt  
<http://www.uniprot.org/uniprot/Q9NWZ3>

## **Sources for This Summary**

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